326 MULTIPLE PTERYGIUM SYNDROME IN IDENTICAL TWINS. Carlos Duran S., Roger L. Ladda, Keith H. Marks. The Penn St Univ Coll Med, MS Hershey Med Ctr, Dept Ped, Hershey, PA.

Multiple pterygium syndrome is a relatively rare disorder characterized by pterygia of the neck, axilla, digits, antecubital, popliteal and intercrural areas. Affected individuals may also have multiple congenital contractures and the diagnosis of arthrogryposis is frequently the presenting complaint. We describe monochorionic diamniotic female twins with multiple pterygium syndrome. The twins were born at 32 weeks' gestation to a 19-year-old G2 P1001. The suspicion of cystic hygroma in one twin was raised during ultrasound screening of the pregnancy at 22 weeks' gestation and the cystic mass was found in Twin B at delivery; it resolved in the first few days of life. Each newborn presented with flexion deformities of the upper and lower extremities, pterygia of the neck, upper arms, antecubital and popliteal regions. The most prominent pterygia involved the upper extremities and neck. Each had angular facies with micrognathia. Twin A had left sided talipes equino varus with right side vertical talus. Twin B has bilateral talipes equino varus. Twin A had a cleft of the soft palate. Twin B had Tetralogy of Fallot requiring a modified Blalock Taussig shunt in the newborn period. Chromosomal studies showed normal female karyotypes in both. Although most cases are sporadic, some appear to be familial and may follow an autosomal recessive pattern of inheritance. Cardiac anomalies in this condition are infrequently reported. Twin A responded well to treatment and was discharged to the referring hospital. Twin B remains in our hospital and still requires ventilatory assistance. Affected identical twins have not previously been reported. This diagnosis should always be considered on a newborn with pterygia.

327 HEMIFACIAL HYPOPLASIA, HYPOPIGMENTATION, AND DIGITAL ANOMALY SYNDROME, DJ Eteson, RD Clark, H Kawamoto, and Y Setoguchi, (Sponsored by LJ Shapiro) Harbor-UCLA Med Ctr, Dept Peds, Torrance, CA, and UCLA, Depts Peds & Plastic Surg, Los Angeles, CA.

We report a unique syndrome of craniofacial anomalies, hypopigmentation and digital abnormalities in 2 unrelated children. Patient 1, a 5 y.o. male of normal intelligence, has right facial hypoplasia, coloboma, cupped ear, sensorineural hearing loss, microdontia, hypodontia, and focal areas of scalp alopecia. There is linear hypopigmentation on the lower lip, chest, right arm, hand and leg. The right 2nd metatarsal is short. Growth and development were normal. Patient 2 is a 7 y.o. female with facial features identical to patient 1 including right facial hypoplasia, eyelid coloboma, focal scalp alopecia and microdontia. Linear and swirled areas of hypopigmented skin are present on her left chest, shoulder and right right leg. The right 2nd metatarsal is short and there is postaxial polysyndactyly. This patient also has imperforate anus and a double collecting system. Intelligence is normal.

Chromosome analysis of peripheral blood and normally pigmented and depigmented skin from both patients was normal, ruling out mosaicism. We propose that this new syndrome may involve a defect in neural crest cell proliferation or migration causing both the facial hypoplasia and pigmentary abnormalities.

328 POSTNATAL PLACIOCEPHALY: PATHOGENESIS AND THERAPY IN 40 CASES (1981-86). John M. Graham, Jr., Catherine E. Charman, Robert Chaisson, (Spon. by R.Z. Klein). Dartmouth Medical School, Dartmouth-Hitchcock Medical Center, Dept. Maternal & Child Health, Dept. of Surgery, (Section of Orthopaedics), Hanover NH 03756

We report 40 cases of postnatal plagiocephaly seen over a 6 yr period (1981-86). Twenty cases had mild to moderate plagiocephaly (10 males, 10 females) due to muscular torticollis and responded to physical therapy and repositioning within the first few months of life. In 3 instances, a head positioning device was helpful; in 2 instances where no consistent therapy was attempted the cranial deformity persisted. An additional 9 cases had severe plagiocephaly due to persistent muscular torticollis (8 males, 1 female). When the above measures failed to correct cranial symmetry by 5 to 6 months, helmet therapy was instituted with excellent results. Another 5 cases (3 males, 2 females), had marked occipital flattening from prolonged use of an Infant Seat; 3 improved with repositioning alone, while 2 with associated muscular torticollis required either head positioning device or helmet. Four children with medical problems resulting in prolonged recumbency (CNS abnormalities, intestinal atresia, TE fistula) and associated mild torticollis, responded to simple repositioning. Two other children with unremitting torticollis due to Klippel-Feil anomaly required helmet therapy. Neck stretching exercises and repositioning were most effective in correcting plagiocephaly when instituted early in the first year of life. When torticollis was severe (more common in males) or due to underlying vertebral anomalies, helmet therapy was effective.

†329 PROSPECTIVE MSAFP SCREENING FOR DOWN SYNDROME: THE BAYLOR EXPERIENCE. Frank Greenberg, Judy Garza, Barbara Weyland, Elliot Alpert, Esmie Rose, (Sponsored by Arthur L. Beaudet), Baylor College of Medicine, Institute for Molecular Genetics and Departme of Medicine, Houston, Texas.

After the initial reports of the association of fetal trisomy 21 with low maternal serum alpha fetoprotein (MSAFP) levels in retrospective studies in 1984, we began using low MSAFP levels as a means of screening for an increased risk of Down syndrome using reagents from Clinical Assays. In 1985, a total of 4929 women were screened, of whom 312 (6.3%) had an initial low MSAFP levels. Of these women, 217 (70%) had levels below the level of reliability of the assay and all but 33 had normal levels on repeat 1-2 weeks later. None of these women had a fetus or infant with trisomy 21 by amniocentesis or at birth. Ninety-five women (1.9%) had levels below half the median. Of these women, 12 had persistently low levels on repeat. A total of 21 amniocenteses were done in this group. Three fetuses with trisomy 21 and one fetus with trisomy 18 were detected, a rate of 7.1% fetal trisomy among those women who had amniocentesis for low MSAFP levels. No cases of trisomy 21 were missed. Thus far in 1986, over 10,000 women have been screened. The rate of fetal trisomy in amniotic fluid is about 3%. One infant with trisomy 21 is known to have been missed.

330 AN OPHTHALMOLOGICAL STUDY OF PATIENTS WITH WILLIAMS SYNDROME. Frank Greenberg, Richard A. Lewis, (Sponsored by Arthur L. Beaudet), Baylor College of Medicine, Institute for Molecular Genetics, Department of Pediatrics Department of Ophthalmology, Houston, Texas.

Between July, 1982 and December 1986, 36 patients with Williams syndrome were evaluated. The mean age at examination was 8.2 years with a range of 0.5-43 years. There were 30 whites, 2 blacks, 2 Hispanics, and 1 Asian. There was a tendency toward ocular hypotelorism with short palpebral fissures; the mean inner canthal distance was 38% for age, while the outer canthal distance was 27%. Of the white patients, 23 had blue irides, 6 hazel and 1 brown. All 6 non-white pateitns had brown irides. All patients with brown irides and, in addition, six patients with blue and three patients with hazel irides did not have stellate iris patterns. Thirty-one percent of the patients had strabismus, similar to previous observations. Three types of optic disc and vascular changes not previously described in the syndrome were noted. Hypermetropic discs were noted in 13 patients. Seventeen patients had simplex vertical branching and three had situs inversus vasorum. Although the latter two findings are considered normal variants, they seem more common in Williams syndrome patients. Accentuated retinal vascular tortuosity has been previously reported, but was not present in any of our patients. In addition, no patients had evidence of ocular manifestations of hypercalcemia. Because of the high incidence of ocular manifestations in Williams syndrome, we recommend ophthalmologic evaluation of all such patients.

331 LISCH NODULES IN AN UNSELECTED POPULATION: PREVALENCE AND USEFULNESS AS INDICATION OF NEUROFIBROMATOSIS. Carol L. Greene, Wendy S. Male, Shelley M. Coleman, Martin E. Ohrlick and Robert A. Gordon (Spon. by Emmanuel Shapira). Tulane Univ Sch of Med, Human Genet Program and Dept of Ophth, New Orleans.

Lisch nodules (LNs), which are found in the iris in neurofibromatosis (NF), were discovered in the otherwise unremarkable parents of two unrelated children with NF. While LNs have not been reported in the absence of NF, there are no studies which test the hypothesis that LNs are pathognomonic for NF. Need for accurate counseling of our families prompted a study to determine the frequency of LNs in the general population. All patients in two ophthalmology clinics were examined by slit lamp before dilation for the presence of LNs. Those with iris trauma or surgery were excluded. In the preliminary stage of the study, 600 patients have been screened and two individuals discovered with one or more LNs. The first is a woman with one small LN without evidence of NF by examination or family history. The second is a man with multiple cafe-au-laits and a seizure disorder, in whom a diagnosis of NF was considered following discovery of LNs and proven by biopsy of a skin tumor.

We suggest that LNs may be found in a limited number of individuals without NF. Nevertheless, we recommend examination for LNs in the ophthalmology clinic as a useful screening tool for NF.